Landmark Seqfirst-neo Study Showcases the Significant Impact of Genomic Testing in the NICU, Revealing Gaps in Current Protocols

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Findings published in the *American Journal of Human Genetics* reveals widespread use of rapid genome sequencing (rGS) demonstrates that more critically ill infants should be tested to reduce missed genetic conditions and promote equitable care for infants at their most critical stage

At least 60% of level IV NICU infants should be receiving rGS

GAITHERSBURG, Md.--(BUSINESS WIRE)--Feb. 24, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced that the *American Journal of Human Genetics* published peer-reviewed research from the Seqfirst-neo study conducted in partnership with Seattle Children's and the University of Washington. Seqfirst-neo is a pioneering study focused on the application of rGS in NICU settings to improve access to a genetic diagnosis overall and specifically in underserved communities to reduce missed diagnoses and improve clinical outcomes for patients.

Seqfirst-neo is the first study to use exclusion, rather than inclusion, criteria for which infants should receive genomic testing in the NICU, setting a new standard of care by enabling neonatologists to more easily identify patients to receive testing, and expanding access to patients who previously would not have been offered testing. Infants were eligible to receive rGS unless their clinical findings were fully explained by birth/physical trauma, complications of prematurity, infection or a pre-existing precise genetic diagnosis (PrGD). Findings from the study show that applying simple exclusion criteria significantly increase the number of infants in the NICU receiving a diagnosis, shorten the time to diagnosis, and drove more equitable access for diverse populations who otherwise would have not received testing. The findings further prove that expanding access to genetic testing dramatically increases the rate of a PrGD, enhances healthcare equity and reduces missed diagnoses.

"There is a critical gap in our current approach to neonatal care – too many critically ill infants and newborns, particularly from underrepresented populations are not being offered genetic testing leading to missed diagnoses and opportunities for precision care," said Mike Bamshad, MD, FACMG, Professor of Pediatrics at the University of Washington School of Medicine and Clinical Genetics Division Chief at Seattle Children's. "Our findings showcase that by shifting today's standard of care to an exclusion-based model for genomic testing, we can significantly expand access, improve health outcomes, and ensure that more families receive the answers they need at the most critical times."

"Seqfirst-neo's findings have had a major impact on the way we deliver genetics services in the inpatient setting. Since implementing exclusion-based identification, we are diagnosing more infants than ever before with genetic conditions in the Seattle Children's Hospital NICU," said Tara Wenger, MD, PhD, FACMG, Professor of Pediatrics at the University of Washington School of Medicine and Associate Medical Director, Inpatient Service at Seattle Children's. "By offering genetic testing at the first unexplained issue in critically ill newborns, we have an opportunity to make diagnoses before they are old enough to experience many of the complications of their genetic disorder. This allows neonatologists and other health care providers the opportunity to introduce the most precise treatments as early as possible."

Seqfirst-neo's findings suggest that at least **60% of Level IV NICU infants should be receiving rGS**. With approximately **400,000 newborn admissions annually across 800 U.S. NICUs**, tens of thousands of infants with genetic conditions are likely being undiagnosed due to lack of access to testing.

Additional findings from the study:

The study evaluated 408 infants in the NICU, of whom 59% met eligibility criteria for rGS. Of those eligible, 126 infants were enrolled in the interventional group (IG) and received rGS, while others followed current diagnostic workflow protocols.

- Nearly half (49.2%) of infants in the IG received a precise genetic diagnosis (PrGD) — an unexpectedly high yield despite broad testing criteria, compared to conventional care (9.7%). **The odds of receiving a PrGD was nine times higher** in the IG compared to conventional care.
- **42% of diagnosed infants would have been missed using conventional NICU protocols (69% of whom were non-white)**, highlighting the limitations of current diagnostic approaches and the correlated inequity of care.
- **24% of diagnosed infants were not suspected of having a genetic condition** based on EMR review and would not have been offered testing under standard care protocols.
- By using simple, broad exclusion criteria, the diagnostic yield and access to testing were comparable across racial groups, with significantly more non-white and Black infants receiving a PrGD than through conventional care, effectively mitigating racial disparities.
- Access to a PrGD led to a **change in clinical management for nearly 97% of diagnosed infants**, influencing medical consultations, additional testing, medication adjustments and family health implications.

“With today’s reliable and rapid genomic technology, we can now deliver answers in days. More importantly, we finally have clear, simple criteria to guide neonatologists in determining which infants should receive genomic screening in the NICU,” said Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. “There is an urgent need to make rapid genome sequencing (rGS) a standard part of NICU care to prevent missed diagnoses and ensure every infant gets the critical care they deserve. By expanding access, we can dramatically improve early detection of genetic conditions, enabling timely interventions and driving more equitable care for all infants.”

GeneDx is a proud collaborator of SeqFirst and their on-going research, as the clinical lab behind both the Seqfirst-neo and SeqFirst Developmental Differences (DDi) projects. Seqfirst-neo focuses on evaluating whether a genotype-driven workflow, using broad and simple exclusion criteria for eligibility, enhances access to a PrGD in critically ill newborns. SeqFirst DDi aims to promote early and equitable access to PrGD for children with atypical development by offering genome sequencing at the time of presentation, rather than after a traditional staged evaluation and testing process.

Accelerating care in the NICU:

GeneDx recently announced ultraRapid Whole Genome Sequencing, offering accelerated and comprehensive genomic insights for neonatal and pediatric patients in the NICU and PICU in as soon as 48 hours. As part of the company’s commitment to increasing access to genomic testing, GeneDx also recently integrated with Epic Aura, enabling health systems to deliver fast and accurate genetic diagnoses to patients and accelerate the path to treatment. By driving collaborative research that informs utilization protocols, continuing to decrease turnaround times, and expanding access to testing, more patients can benefit from GeneDx’s genomic insights of over 750,000 exomes and genomes and increase the likelihood of obtaining definitive diagnoses.

About GeneDx:

At GeneDx (Nasdaq: WGS), we believe that everyone deserves personalized, targeted medical care—and that it all begins with a genetic diagnosis. Fueled by one of the world’s largest rare disease data sets, our industry-leading exome and genome tests translate complex genomic data into clinical answers that unlock personalized health plans, accelerate drug discovery, and improve health system efficiencies. For more information, please visit genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

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